

Public involvement in global genomics research: A scoping review

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Abstract

Public involvement in research occurs when the public, patients or research participants are actively contributing to the research process. Public involvement has been acknowledged as a key priority for many prominent human genomics research initiatives worldwide. However, to date, there has been no detailed analysis or review of the features, methods and impacts of public involvement occurring in human genomics research projects worldwide. Here, we review the reported public involvement in 96 human genomics projects (initiatives), based on a database of initiatives hosted by the Global Alliance for Genomics and Health, according to information reported on public domain websites. To conduct the scoping review, we applied a structured categorization of criteria to all information extracted from the search. We found that only a third of all initiatives reported public involvement in any capacity (32/96, 33%). In those reporting public involvement, we found considerable variation in both the methods and tasks of involvement. Some noteworthy initiatives reported diverse and comprehensive ways of involving the public, occurring through different stages of the research project cycle. Three notable initiatives reported a total of eight distinct impacts as a result of involving people. Our findings suggest there would be intrinsic value in having more public involvement occur in human genomics research worldwide. We also suggest more systematic ways of reporting and evaluating involvement would be highly beneficial, to help develop best practices.

Introduction

In human genomics, there is a growing need to increase the involvement of the public in research and policy development, as this has been identified as a crucial aspect of responsible research practice^{1,2}. The concept of ‘public involvement’ in research is defined as research that is carried out ‘with’ people rather than ‘on’ them³. Public involvement can also be defined as when the public, patients or research participants actively contribute to the research or policy development process⁴.

The number of people involved in genomics research is predicted to grow substantially in coming years^{5,6}. By 2025, it is estimated that nearly 2 billion people worldwide will have had their DNA sequenced, creating a global imperative for responsible and effective public involvement in research¹⁰. Many high-profile genomics research initiatives have already made public statements about the importance of involving people, with some governments positioning public involvement as a democratic right⁷⁻⁹.

The benefits of involving the public in research are wide-ranging. They include improving trust and public influence over research^{1,7,10}; ensuring that research is conducted in an ethical, accessible and transparent manner; and ensuring that research reflects the balance and diversity of priorities within populations^{11,12}. However, with the growing interest and importance of large-scale human genomics initiatives worldwide, there has been limited research into how the public are currently being involved. There has also been no assessment of the resulting impacts and benefits, including genomics initiatives that have involved the public.

While involving the public in other types of health and medical research has been the subject of previous systematic reviews¹³⁻¹⁵, comparable reviews have not been published in human genomics. Many of the existing reviews on other areas of medical research conclude that reports of involvement activities are inconsistent or under-reported¹⁵⁻¹⁹ and that the precise ways in which people are involved in medical research are not well reported, including any impacts from involving people^{10,14,16}.

Our review provides a summary of currently reported public involvement in 96 global human genomics projects, listed on a database managed by the Global Alliance for Genomics and Health (GA4GH), a recently formed international organization seeking to enable responsible genomics data sharing within a human rights framework²⁰. The list provides a representation of the current landscape of human genomics research worldwide, and a snapshot of contemporary practice with regards to public involvement in genomics research.

This scoping review provides a new perspective by exploring how these genomics initiatives have conducted and reported public involvement to date, including any impacts, facilitators and barriers of involvement. The intention is that resulting data will help inform future directions for integrating

public involvement into genomics research and policy development, and inform the development of ways of routinely reporting and evaluating any involvement.

Methods

Source

Using a list of human genomics research projects (“initiatives”) from a database hosted by the GA4GH (see Supplementary Materials ‘Table 1’), we systematically searched public domain websites for information reported on involving the public in research. The database was curated by GA4GH staff, last verified August 2016, and contains information about the type of the genomics research initiative (i.e. consortium, data-sharing initiative, organisation(s), repository or research project), the type of data gathered (i.e. whole-genome or whole-exome sequencing), the geographical scope of the initiative, number of participants (cohort size), relevant disease areas, and the public domain URL of the website for the organisation or initiative (as some ‘initiatives’ involve a number of organisations). The scoping review methodology can be summarized in three stages (see ‘Figure 1: Scoping review study overview and results summary’ for overview):

Stage 1 – Defining “involvement” and the search strategy

We developed a criteria to define ‘involvement’ based on the International Association for Public Participation’s participation spectrum and other studies^{4,7,21,22}. This included reports of ‘consultation’, ‘involvement’, ‘collaboration’ and ‘empowerment’²³. Involving people in genomics research was defined as the ‘active involvement’ in shaping and guiding research, rather than only providing data^{17,23,24}. We defined specific tasks related to involvement at different stages of the research cycle³, such as the sharing of views to influence research, or co-creating the research^{19,25,26}. ‘Consequential’ involvement meant involvement contributing to the research process, as distinct from involvement which is ignored or not incorporated^{27–29}. We could not always determine whether involvement was consequential based on the available information, so an assumption was made that all methods reported resulted were ‘consequential’.

Stage 2 – Searching websites (data extraction)

Public domain websites of all the initiatives in the GA4GH database were searched for reports of involvement and associated impacts. The date range for website searching and data extraction was 16th August to 28th November 2017. The exact text from the URLs where data was extracted from was collected to allow reanalysis, with all relevant URLs archived using an online archive service to preserve the content and the date of extraction³⁰.

We used search engine operators to systematically scan the text of each public domain website for relevant phrases, including all grammatical variations of the words used (for example, deriving ‘involvement’, ‘involves’, ‘involved’ and ‘involving’ from the root word ‘involve’). Grammatical variations of specific phrases (denoted by inverted commas) were generated using tables to systematically create a series of search strings for each domain. For example, this search string returned 4 results:

site:www.ukbiobank.ac.uk/ “public involvement” OR “involves public” OR “public involved”
OR “involving the public” OR “involve public”

Reports of involvement were assessed by a member of the research team (JN), then independently assessed by an additional member of the research team, with a random sample assessed by a third investigator (PL). Any disagreements between the team on the data included was discussed until a consensus was reached. Informed by previous reviews, the search terms for the concept of involvement were; “engagement”, “involvement” and “partnership”^{22,31–34}. The search terms to describe the people involved were; ‘citizen(s)’, ‘community’, ‘consumer(s)’, ‘lay’, ‘patient(s)’, ‘public’, ‘stakeholder(s)’ and ‘user(s)’.

In addition to using a standard list of terms, adaptive (context dependent) search terms were sometimes required when searching domains where terms were specific to the region or initiative. Adaptive search terms were; ‘advocate(s)’, ‘carer(s)’, ‘civil society’, ‘client(s)’³⁵, ‘customer(s)’³⁵, ‘group(s)’, ‘participant(s)’, ‘payer(s)’, ‘population(s)’²⁹, ‘PPI’ (an acronym commonly used in the UK which stands for ‘patient and public involvement’), ‘residents’ (geographical grouping)³⁶, ‘representative(s)’, ‘taxpayer(s)’ and ‘volunteer(s)’. For more details on search method, see Supplementary Materials ‘Table 2’.

Stage 3 – Defining inclusion/exclusion criteria, data synthesis and analysis

Defining the inclusion and exclusion criteria was an iterative process informed by published scoping review methodologies^{37,38}. Initiatives reporting no involvement were excluded from further analysis. Initiatives were categorised as ‘no involvement’ if the context of words such as ‘participation’ were used to describe ‘research participants’ (research subjects) only, rather than aligning with the concept of involvement already articulated^{4,39}. Reported impacts were excluded if they were phrased as anticipated future impacts (using terms such as ‘we expect’), rather than reporting real results. Initiatives reporting ‘data sharing’ as the only type of involvement were also excluded. Initiatives reporting any other type of involvement, according to our definition, were included and proceeded to data extraction (structured categorization of extracted search data).

Extracted data was categorized (data synthesis) based on the following types of information; **a)** the *method* of involvement (*how* people were involved)²⁴; **b)** the *tasks* they were involved in (what people *did*)²⁴; **c)** the *stage* of the research (using an expanded version of an existing framework¹⁵, informed by INVOLVE)⁴⁰; **d)** *who* was involved, for example ‘research participants’, ‘patients’ and ‘public’ (informed by the Concannon ‘7Ps Framework’ taxonomy)¹⁶; **e)** *reported facilitators or barriers* of involvement; and **f)** publicly-reported *impacts* (informed by section 7 and 8 of the GRIPP2 framework)^{16,24}.

As there is currently no standardised way to report and group methods of involving people or descriptions of people involved^{24,29}, grouping was informed by methods of previous reviews (for example, grouping similar methods of involving people²⁴) and by using previously established nomenclature^{26,33,39}. The initial grouping (JN) was reviewed by other authors (PL). While previous reviews have used frameworks to label the ‘roles’, ‘degrees’ or ‘levels’ of involvement or ‘control’^{19,24}, we did not use these frameworks as they require subjective judgements to be made, often with insufficient data^{26,41–43}.

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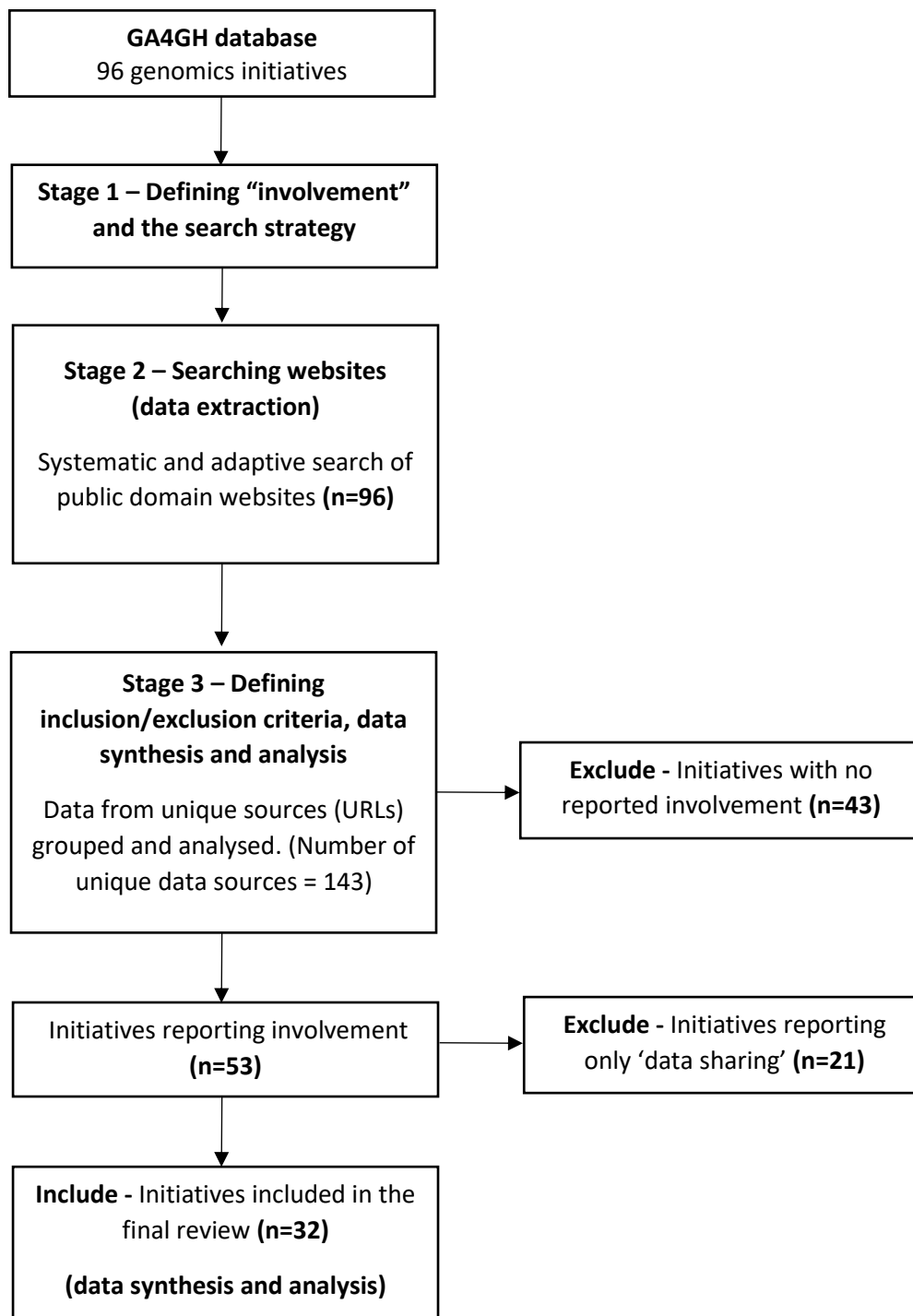


Figure 1: Scoping review study overview and results summary

Results

Of the 96 initiatives searched, based on our criteria, only a third reported involving people in some capacity (32/96, 33%) (Table 1). These 32 initiatives were included in the final analysis (data synthesis).

Reported methods of involvement

The reported methods of involving people were organised into categories, shown below in **bold**, with the number of total initiatives reporting each method shown in brackets:

- **Citizen science** (n=2) - people involved beyond data collection, research design or data analysis, towards co-creation across all aspects of the scientific process⁴⁴;
- **Consultation** (n=4) - an organised consultation or dialogue process;
- **Formal discussion** (n=8) - formalised 'focus groups', forums or interview structures;
- **Formal groups** (n=20) - a working group or committee (including ethics and data access committees, 'scientific advisory groups' and 'steering groups');
- **Generic involvement** (n=11) - informal, such as meetings, 'partnership', or an unspecified method;
- **Newsletters** (n=2) - or mailing lists;
- **Online tools** (n=7) - websites, social media, or online community hosting;
- **Public events** (n=13) - with discussion - including initiatives hosting public debates, workshops, discussion spaces or conferences;
- **Surveys** (n=10) - including questionnaires; and
- **Other** (n=7) - methods not described by other categories.

Some initiatives reported using multiple methods to involve people. Reports of involving people also showed that some methods, for example 'formal discussion', can use different modes of communication, including face to face, online (for example, 'massive open online courses'), or a combination of the two.

Table 1: Summary of G44GH initiatives reporting public involvement

Initiatives from a database provided by the Global Alliance for Genomics and Health were searched for reports of public involvement (based on public domain websites). Each initiative has been assigned an ID number. The type (method) of involvement was categorized using specific criteria.

Name of Initiative/Organization	ID	Geographic Region (cohort size)	Reported methods of involving people
100k Wellness Project	1	North America (100000)	Online tools, Other
Australian Genomics Health Alliance (AGHA)	8	Australia (1800)	Formal groups, Other, Public events
Biobanking and Biomolecular resources Research Infrastructure (BBMRI)	11	Europe (N/A)	Formal discussion formats, Public events
Cancer MoonShot 2020	16	North America (20000)	Generic involvement
Clinical Sequencing Exploratory Research (CSER)	21	North America (6000)	Generic involvement
DECIPHER	24	International (21475)	Formal groups
East London Genes & Health	26	Europe (100000)	Formal groups, Generic involvement
Electronic Medical Records and Genomics (eMERGE)	27	North America (55028)	Surveys
ELIXIR	28	Europe (N/A)	Consultation, Formal groups, Public events
France Genomic Medicine 2025	33	Europe (N/A)	Consultation, Generic involvement
Genome in a Bottle	35	International (N/A)	Generic involvement, Public events
Genomics England	37	Europe (100000)	Consultation, Formal discussion formats, Formal groups, Generic involvement, Other, Public events, Surveys
H3Africa	41	Africa (60000)	Formal discussion formats, Generic involvement
Implementing Genomics in Practice (IGNITE)	44	North America (73000)	Formal groups, Public events
International Rare Diseases Research Consortium (IRDiRC)	50	International (N/A)	Formal groups, Generic involvement, Other, Public events
Kaiser Permanente Research Program on Genes, Environment, and Health (RPGEH)	52	North America (500000)	Formal groups
Kaviar	53	North America (N/A)	Formal groups

Matchmaker Exchange	57	International (N/A)	<i>Formal groups, Online tools</i>
MSSNG	60	North America (10000)	<i>Formal groups</i>
MyCode Community Health Initiative	62	North America (250000)	<i>Formal groups</i>
MyGene2	63	International (500)	<i>Online tools</i>
openSNP	65	Europe (2500)	<i>Citizen science, Online tools, Surveys</i>
Precision Medicine Initiative / ‘All of Us’	69	North America (1000000)	<i>Citizen science, Formal groups, Formal discussion formats, Generic involvement, Online tools, Other, Public events, Surveys</i>
Public Population Project in Genomics and Society (P3G)	72	International (N/A)	<i>Formal groups, Online tools, Public events, Surveys</i>
Qatar Genome Project	73	Asia (1161)	<i>Surveys</i>
RD-Connect	74	Europe (2500)	<i>Formal discussion formats, Formal groups, Generic involvement, Newsletters, Online tools, Surveys</i>
The Clinical Genome Resource (ClinGen)	84	North America (N/A)	<i>Formal groups, Other</i>
Tohoku Medical Megabank Project	86	Asia (150000)	<i>Formal discussion formats, Public events, Surveys</i>
Transforming Genetic Medicine Initiative (TGMI)	88	Europe (N/A)	<i>Public events</i>
UK Biobank	92	Europe (500000)	<i>Consultation, Formal discussion formats, Formal groups, Generic involvement, Newsletters, Other, Public events, Surveys</i>
Undiagnosed Diseases Network (UDN)	94	North America (8000)	<i>Formal groups</i>
Vanderbilt’s BioVU	96	North America (215000)	<i>Formal groups, Public events</i>

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Figure 2 summarizes overall findings from data synthesis. There was variability in the methods and tasks of involvement reported. This supports previous findings that involvement in biomedical research is diverse, varied and described using different language⁴⁵.

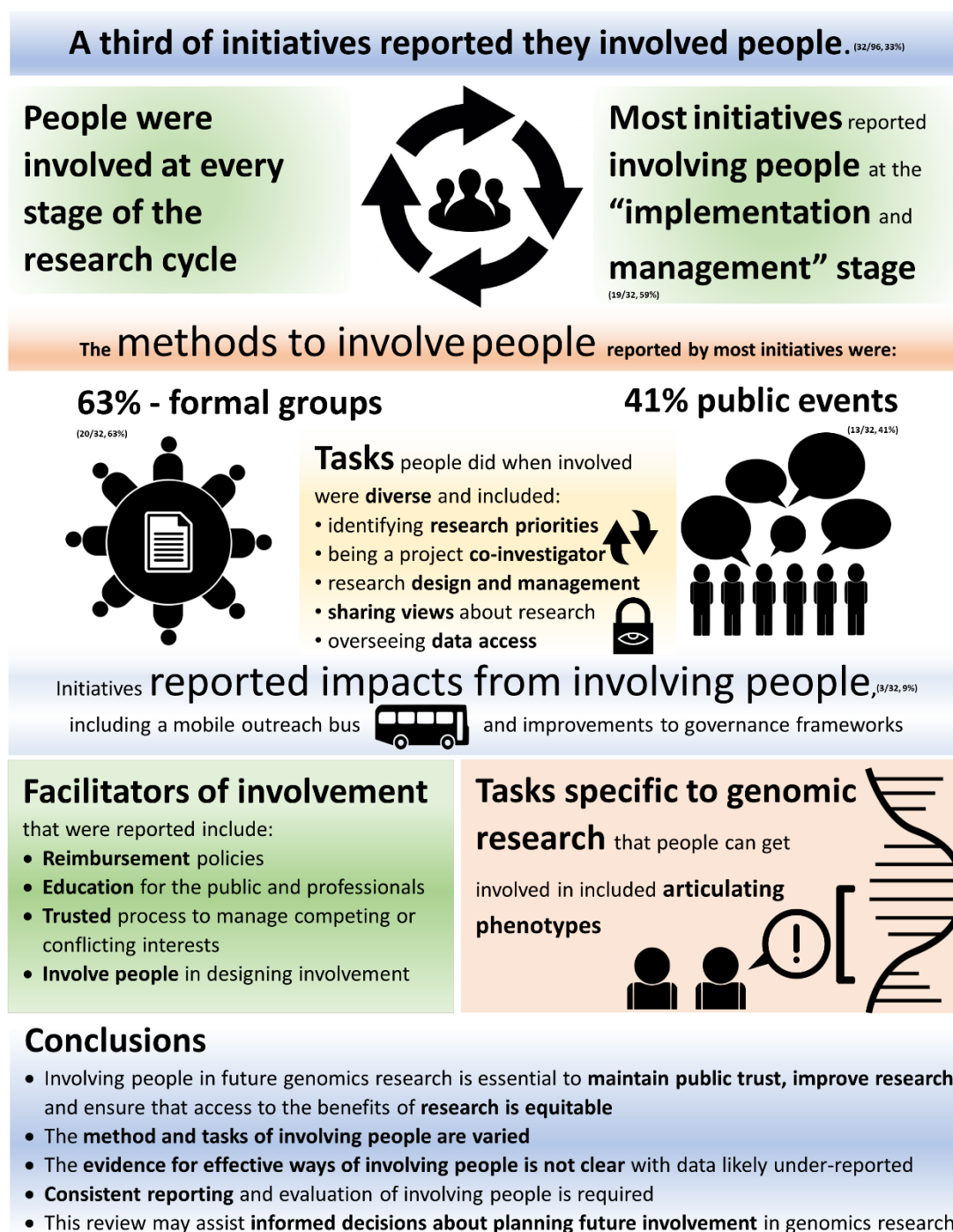


Figure 2 – Summary of results

Reported tasks of involvement

The tasks people were involved in (what people did when they were involved) were diverse. Tasks included; identifying research priorities related to people with specific diseases; communicating priorities to scientists, clinicians and health policy makers, [IDs 11, 37, 50, 74]; designing or improving how people will be involved in the research [IDs 41, 50]; educating professionals involved in the research [ID 8]; developing workshops and conferences [IDs 44, 94], offering culturally appropriate information about research to people in community groups [ID 37]; providing feedback on the cultural and linguistic appropriateness of public domain research documents [ID 96]; and translating information into 'lay' language [ID 92]. Tasks also involved sharing views and perspectives about multiple aspects of research projects [ID 37, 92, 96]; articulating phenotypes [ID 65]; and being a project co-investigator [ID 21].

Some initiatives reported involving people in the task of giving feedback and sharing views and perspectives about the 'acceptability' of specific aspects of the research design. For example, research management, governance [IDs 27, 41, 92, 72], accountability, planning, policy, protocols, data access and data use [IDs 37, 74, 84, 92], consent, re-contact, withdrawal, confidentiality, benefit sharing, project closure and recruitment [IDs 37, 62, 74, 92]. A number of initiatives also involved people in the task of sharing views and perspectives on issues of perceived social and ethical importance (including being told about potentially serious incidental findings) [IDs 37, 74, 96], or to scrutinise a project to ensure it aligned with public interest [ID 11].

Reported stages of involvement

Most reports of involvement were at the 'implementation and management' stage of research (19/32, 59%), followed by 'dissemination' (12/32, 38%), 'evaluation' and 'study design' (both 9/32, 28%) and 'data analysis' (8/32, 25%). The stage with the lowest number of initiatives reporting involvement was 'funding' (1/32, 3%) with the next lowest being 'identifying topics' and 'prioritisation' (both 4/32, 13%). Four initiatives reported involving people at every stage of research [IDs 21, 50, 69, 74].

Reported impacts of involvement

Nearly 10% of the initiatives reporting involvement also reported impacts of the involvement (3/32, 9.4%). The method with the most reported impacts was 'public events' (4/8, 50%), followed jointly by 'formal discussion formats' and 'surveys' (2/8, 25%). Actions taken as a result of involving people included the creation of a mobile outreach bus [ID 37]; improvements to ethical and governance frameworks [ID 92]; and improved participant information and consent documents [ID 37]. Three initiatives reported a total of eight distinct impacts as a direct result of involving people. Some impacts were reported as being a result of using a combination of methods.

222 *Reported facilitators and barriers to involvement*

223 A number of specific facilitators of involvement were reported, including: reimbursement policies [ID
224 21], with people involved paid for their time [IDs 92, 94], travel [IDs 74, 94] accommodation [ID 74]
225 and expenses [IDs 74, 92]; education and learning opportunities for the general public [IDs 1, 11, 41],
226 ensuring people involved are informed and can make informed decisions [ID 11]; education for health
227 professionals [IDs 41, 50], providing opportunities to learn about how to involve people [IDs 41, 50],
228 and governance which is trusted by all stakeholders to be able to manage real or perceived competing
229 or conflicting interests [ID 50]. The only barrier reported was limited venue size, which inhibited the
230 number of people involved [ID 92]. This also implies a limited budget, which is an important but
231 likely under-reported implicit limitation on all involvement methods.

232

Discussion

This review provides an overview of reported public involvement occurring in prominent human genomics projects worldwide, during a period of rapid growth for genomics research. We identified significant variability in the way in which involvement is reported. This variation of reported involvement suggests diversity in both the ways people are being involved in genomics, and in the varied and emergent language used to report and describe involvement, consistent with other areas of biomedical research^{7,46}. While there are similarities with the principles of involvement in other kinds of biomedical research, this review has identified three different tasks not found in other reviews⁴⁵.

Because the results from this review suggest there is currently no standardized way of reporting and therefore evaluating how people are involved, there is a risk that best-practice will be hard to define or even absent in future evidence reviews. This has implications, as the number of people involved in genomic research is predicted to grow exponentially. Without a standardized framework to report and transparently evaluate ways people are involved, it will be difficult to create an evidence base to inform best-practice.

While a third of initiatives reported involvement, a majority of projects did not (64/96, 66%). Some prominent initiatives involving the genetic analysis of thousands of people did not refer to public involvement in any way. This is somewhat concerning given that involving the public has been identified as a crucial aspect of responsible research practice in genomics¹. Whilst we acknowledge the probable under-reporting of involvement activities on public-domain websites, we argue public involvement in human genomics needs to increase.

Findings from this review also suggest it is best-practice to involve multiple stakeholders (including the public) in designing how people will be involved in research (co-design of involvement plan), to involve the public throughout the lifetime of a project in certain tasks (such as overseeing data access) and to evaluate the involvement with both qualitative and quantitative data.

Involving people in planning involvement may improve how appropriate, effective, efficient and equitable it is. Involving people in the design of planned methods of involvement by identifying what is considered 'good practice' was reported by H3Africa [ID 41] and the International Rare Diseases Research Consortium (IRDiRC), and reported as a facilitator of involvement by the IRDiRC [ID 50]. The IRDiRC [ID 50] also reported both qualitative and quantitative data should be used to evaluate involvement, although there is currently no way to systematically collect and analyse such activity⁴⁸.

If involvement is more effective when the public are invited to help plan it, standardised reporting and evaluation will help make informed decisions at every stage of involvement from co-design through to evaluation.

Implications for policy and practice

With the impact of some genomics research data likely to be measured in decades, some of the initiatives offer a useful insight into planning and funding sustainable (long-term) involvement for the entirety of an initiative's lifetime⁸. For example, Genomics England [ID 37] and the UK Biobank [ID92], as exemplars, both reported multiple ways of involving people, at different stages of the research cycle, conducted over a number of years. Other initiatives, such as the International Rare Diseases Research Consortium (IRDiRC) [ID 50] and the Public Population Project in Genomics and Society (P3G) [ID 72], also publicly stated the importance of planning sustainable involvement over the duration of a project. These initiatives demonstrate a standard of involving people which could eventually be used to inform international best practice.

The IRDiRC also reported that involving people throughout an entire project helped maintain trust by scrutinising and managing competing or conflicting interests [ID 50]. Similarly, the UK Biobank reported that involving people in the ethics and governance should not be one-off and must be ongoing [ID 92]. The method of using 'formal groups' was more common for more complex or ongoing tasks such as overseeing data access, policy development, research management and improving research protocols.

Some initiatives, such as openSNP, reported tasks that were specific to genomics research, such as articulating phenotypes [ID 65]. Involvement in this kind of task might have important implications when working to usefully describe people's subjective lived-experiences across multiple languages, for example, rare diseases and mental health⁴⁹.

Public involvement in articulating phenotypes also suggests that the traditional boundaries between terms such as 'research', 'healthcare', 'patients', 'research participants' and 'the public' may be increasingly challenged by the methodology of future genomic research⁵⁰. Findings from this review show that both 'the public' and 'patients' are already involved in every stage of research, including collecting and analyzing data⁵⁰. Any future standardised reporting of involvement will need to keep pace with the continually evolving language to describe not only what research is, but who is involved and how.

297 *Limitations*

298 While the database hosted by GA4GH includes many of the most prominent human genomics
299 research initiatives worldwide, the database is not exhaustive. There are several known genomics
300 initiatives which involved people that were not part of the database. Therefore, the GA4GH selection
301 cannot be considered entirely systematic or representative. However, it does provide a reasonable
302 indication and snapshot of the current global landscape in human genomics research.

303 Our data collection was limited to self-reported information reported on English language websites
304 only. This likely under-reports the total amount of public involvement occurring. For example, some
305 initiatives may have conducted involvement, and not reported it publicly. This indicates a current lack
306 of standardization or best-practice in reporting involvement activities in human genomics research,
307 which we feel could be improved.

308 Of the public involvement activities reported, we did not systematically follow up reports to confirm
309 they had taken place, or if involvement was ‘consequential’^{27–29}. While this is a limitation of the
310 review, it also reflects the inconsistent and often incomplete ways genomics research initiatives report
311 impacts of involving people. For example, the impact of how involvement influenced research was
312 only reported by three projects - Genomics England [ID 37], the Qatar Genome Project [ID 73] and
313 the UK Biobank [ID 92].

314 A number of reported methods did not provide sufficient information to make a clear decision about
315 how to group a method. For example, many reports of involvement simply referred to a ‘workshop’,
316 ‘meeting’, or other ‘public events’, where people were able to get involved by sharing views and
317 perspectives. As a result there is potentially significant overlap between some methods, which could
318 have been articulated more clearly if more data were available.

319 Reports of ‘data sharing’ were excluded, as they were not considered as public involvement. While
320 sharing data may enable people to be involved in some capacities (for example, in analysing data),
321 data sharing is not necessarily an indicator that people were involved in the analysis of data. The
322 complexity within the term ‘data sharing’ in genomics, and how people can be involved in the
323 analysis and interpretation of data also requires further consideration^{51–53}.

324

325 *Conclusion*

326 Involving people in the future of genomics research is essential to maintain public trust, improve
327 research outcomes, and to ensure that access to the benefits of research is equitable^{1,14,50}. While a third
328 of initiatives reported involving people, only 10% of initiatives reported impacts. The limited
329 reporting of involvement suggests there would be intrinsic value in developing a more systematic

method of both reporting and evaluating how people are involved in human genomics research. Data from such reporting could provide the evidence required to inform future policy around involvement of the public, as human genomics research continues to grow.

Supplementary Material

- ‘GA4GH database and results table’
 - This table combines a database of human genomics research projects hosted by the ‘Global Alliance for Genomics and Health’ with a summary of results from a scoping review of currently reported public involvement reported on public domain websites. The data is organised alphabetically, with organisations reporting involvement listed first.
- ‘Systematic search method’
 - This document describes the search method, including how standard and adaptive were used to search the public domain websites of all the included initiatives in the GA4GH database for reports of involvement and any impacts.

346 **References**

- 347 1. Burton H, Adams M, Bunton R, et al. Developing stakeholder involvement for introducing
348 public health genomics into public policy. *Public Health Genomics*. 2009;12(1):11-19.
349 doi:<https://dx.doi.org/10.1159/000153426>.
- 350 2. Lemke AA, Harris-Wai JN. Stakeholder engagement in policy development: challenges and
351 opportunities for human genomics. 2015. doi:10.1038/gim.2015.8.
- 352 3. National Institute for Health Research. Briefing note eight: Ways that people can be involved
353 in the research cycle. National Institute for Health Research.
354 [http://web.archive.org/web/20170605035051/http://www.invo.org.uk/posttypesresource/where-](http://web.archive.org/web/20170605035051/http://www.invo.org.uk/posttypesresource/where-and-how-to-involve-in-the-research-cycle/)
355 [and-how-to-involve-in-the-research-cycle/](http://web.archive.org/web/20170605035051/http://www.invo.org.uk/posttypesresource/where-and-how-to-involve-in-the-research-cycle/). Published 2017. Accessed June 5, 2017.
- 356 4. National Institute for Health Research. Patient and public involvement in health and social care
357 research. 2014. [https://www.nihr.ac.uk/about-us/CCF/funding/how-we-can-help-you/RDS-](https://www.nihr.ac.uk/about-us/CCF/funding/how-we-can-help-you/RDS-PPI-Handbook-2014-v8-FINAL.pdf)
358 [PPI-Handbook-2014-v8-FINAL.pdf](https://www.nihr.ac.uk/about-us/CCF/funding/how-we-can-help-you/RDS-PPI-Handbook-2014-v8-FINAL.pdf). Accessed February 2, 2018.
- 359 5. Sandhu C, Qureshi A, Emili A. Panomics for Precision Medicine. *Trends Mol Med*.
360 2018;24(1):85-101. doi:10.1016/J.MOLMED.2017.11.001.
- 361 6. Green ED, Rubin EM, Olson M V. The future of DNA sequencing. *Nature*.
362 2017;550(7675):179-181. doi:10.1038/550179a.
- 363 7. Keltz C, Panofsky A. Disentangling public participation in science and biomedicine. *Genome*
364 *Med*. 2014;6(1):8. doi:10.1186/gm525.
- 365 8. Wagner JK, Peltz-Rauchman C, Rahm AK, Johnson CC. Precision engagement: the PMI's
366 success will depend on more than genomes and big data. *Genet Med*. 2016;(October):1-5.
367 doi:10.1038/gim.2016.165.
- 368 9. Sally Davies et al. *Annual Report of the Chief Medical Officer 2016 - "Generation Genome."*;
369 2017.
370 [https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/624628/CMO_a](https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/624628/CMO_annual_report_generation_genome.pdf)
371 [nnual_report_generation_genome.pdf](https://www.gov.uk/government/uploads/system/uploads/attachment_data/file/624628/CMO_annual_report_generation_genome.pdf).
- 372 10. Brett J, Staniszewska S, Mockford C, et al. Mapping the impact of patient and public
373 involvement on health and social care research: a systematic review. *Health Expect*.
374 2014;17(5):637-650. doi:10.1111/j.1369-7625.2012.00795.x.
- 375 11. Crowe S, Fenton M, Hall M, Cowan K, Chalmers I. Patients', clinicians' and the research
376 communities' priorities for treatment research: there is an important mismatch. *Res Involv*
377 *Engagem*. 2015;1(1):2. doi:10.1186/s40900-015-0003-x.
- 378 12. World Health Organisation. Declaration of Alma-Ata.
379 http://www.who.int/publications/almaata_declaration_en.pdf?ua=1. Published 1978. Accessed
380 June 25, 2018.
- 381 13. Domecq JP, Prutsky G, Elraiyah T, et al. Patient engagement in research: a systematic review.
382 *BMC Health Serv Res*. 2014;14(1):89. doi:10.1186/1472-6963-14-89.
- 383 14. Brett J, Staniszewska S, Mockford C, et al. A Systematic Review of the Impact of Patient and
384 Public Involvement on Service Users, Researchers and Communities. *Patient*. 2014;7(4):387-
385 395. doi:10.1007/s40271-014-0065-0.
- 386 15. Shippee ND, Domecq Garces JP, Prutsky Lopez GJ, et al. Patient and service user engagement
387 in research: A systematic review and synthesized framework. *Heal Expect*. 2015;18(5):1151-
388 1166. doi:10.1111/hex.12090.
- 389 16. Staniszewska S, Brett J, Simera I, et al. GRIPP2 reporting checklists: tools to improve

390 reporting of patient and public involvement in research. 2017. doi:10.1186/s40900-017-0062-
391 2.

392 17. Staniszewska S, Adebajo A, Barber R, et al. Developing the evidence base of patient and
393 public involvement in health and social care research: the case for measuring impact. *Int J*
394 *Consum Stud*. 2011;35(6):628-632. doi:10.1111/j.1470-6431.2011.01020.x.

395 18. Staley K, Buckland SA, Hayes H, Tarpey M. “The missing links”: Understanding how context
396 and mechanism influence the impact of public involvement in research. *Heal Expect*.
397 2014;17(6):755-764. doi:10.1111/hex.12017.

398 19. Oliver SR, Rees RW, Clarke-Jones L, et al. A multidimensional conceptual framework for
399 analysing public involvement in health services research. *Heal Expect*. 2008;11(1):72-84.
400 doi:10.1111/j.1369-7625.2007.00476.x.

401 20. Terry SF. The Global Alliance for Genomics & Health. *Genet Test Mol Biomarkers*.
402 2014;18(6):375-376. doi:10.1089/gtmb.2014.1555.

403 21. Concannon TW, Meissner P, Grunbaum JA, et al. A new taxonomy for stakeholder
404 engagement in patient-centered outcomes research. *J Gen Intern Med*. 2012;27(8):985-991.
405 doi:10.1007/s11606-012-2037-1.

406 22. Rogers M, Bethel A, Boddy K. Development and testing of a medline search filter for
407 identifying patient and public involvement in health research. *Health Info Libr J*.
408 2017;34(2):125-133. doi:10.1111/hir.12157.

409 23. International Association for Public Participation. Participation Spectrum.
410 https://www.iap2.org.au/Tenant/C0000004/00000001/files/IAP2_Public_Participation_Spectrum.pdf. Published 2014. Accessed July 12, 2018.

412 24. Oliver S, Clarke-Jones L, Rees R, et al. Involving consumers in research and development
413 agenda setting for the NHS: Developing an evidence-based approach. *Health Technol Assess*
414 *(Rockv)*. 2004;8(15):1-148, III-IV. doi:10.3310/hta8150.

415 25. Woolley JP, McGowan ML, Teare HJA, et al. Citizen science or scientific citizenship?
416 Disentangling the uses of public engagement rhetoric in national research initiatives. *BMC*
417 *Med Ethics*. 2016;17(1):33. doi:10.1186/s12910-016-0117-1.

418 26. Stokes F, Press A, Oliver SR, Liabo K, Stewart R, Rees R. Public involvement in research:
419 making sense of the diversity. *J Health Serv Res Policy*. 2014;20(1):45-51.
420 doi:10.1177/1355819614551848.

421 27. Van Mil A, Hopkins H, Kinsella S. *Potential Uses for Genetic Technologies: Dialogue and*
422 *Engagement Research Conducted on Behalf of the Royal Society Findings Report.*; 2017.
423 [https://royalsociety.org/~media/policy/projects/gene-tech/genetic-technologies-public-](https://royalsociety.org/~media/policy/projects/gene-tech/genetic-technologies-public-dialogue-hvm-full-report.pdf)
424 [dialogue-hvm-full-report.pdf](https://royalsociety.org/~media/policy/projects/gene-tech/genetic-technologies-public-dialogue-hvm-full-report.pdf). Accessed March 19, 2018.

425 28. International Finance Corporation. Stakeholder Engagement : A Good Practice Handbook for
426 Companies Doing Business in Emerging Markets. *Int Financ Corp*. 2007:201.
427 doi:10.1007/s10551-007-9509-y.

428 29. Nilsen ESE, Myrhaug HTH, Johansen M, Oliver SR, Oxman ADA. Methods of consumer
429 involvement in developing healthcare policy and research, clinical practice guidelines and
430 patient information material. In: Nilsen ES, ed. *Cochrane Database of Systematic Reviews*.
431 Chichester, UK: John Wiley & Sons, Ltd; 2006:CD004563.
432 doi:10.1002/14651858.CD004563.pub2.

433 30. Internet Archive. Internet Archive: About IA. <https://archive.org/about/>. Published 2018.
434 Accessed February 2, 2018.

- 435 31. Degeling C, Carter SM, Rychetnik L. Which public and why deliberate? – A scoping review of
436 public deliberation in public health and health policy research. *Soc Sci Med*. 2015;131:114-
437 121. doi:10.1016/j.socscimed.2015.03.009.
- 438 32. Morley RF, Norman G, Golder S, Griffith P. A systematic scoping review of the evidence for
439 consumer involvement in organisations undertaking systematic reviews: focus on Cochrane.
440 *Res Involv Engagem*. 2016;2(1):36. doi:10.1186/s40900-016-0049-4.
- 441 33. Pollock A, Campbell P, Struthers C, et al. Stakeholder involvement in systematic reviews: a
442 protocol for a systematic review of methods, outcomes and effects. *Res Involv Engagem*.
443 2017;3(1):9. doi:10.1186/s40900-017-0060-4.
- 444 34. Lander J, Hainz T, Hirschberg I, Bossert S, Strech D. Do Public Involvement Activities in
445 Biomedical Research and Innovation Recruit Representatively? A Systematic Qualitative
446 Review. *Public Health Genomics*. 2016;19(4):193-202.
447 doi:https://dx.doi.org/10.1159/000444478.
- 448 35. Mitton C, Smith N, Peacock S, Evoy B, Abelson J. Public participation in health care priority
449 setting: A scoping review. *Health Policy (New York)*. 2009;91(3):219-228.
450 doi:10.1016/j.healthpol.2009.01.005.
- 451 36. Collins M. PiiAF The Public Involvement Impact Assessment Framework Guidance.
452 <http://piiaf.org.uk/documents/piiaf-guidance-jan14.pdf>. Published 2014. Accessed October 4,
453 2017.
- 454 37. Levac D, Colquhoun H, O'Brien KK, Brien KKO. Scoping studies: advancing the
455 methodology. *Implement Sci*. 2010;5(1):69. doi:10.1186/1748-5908-5-69.
- 456 38. Arksey H, O'Malley L, Scott SJ, et al. Scoping studies: towards a methodological framework.
457 *Int J Soc Res Methodol*. 2005;8(1):19-32. doi:10.1080/1364557032000119616.
- 458 39. Hill S. Involving the Consumer in Health Research. In: Saks M, Allsop J, eds. *Researching
459 Health: Qualitative, Quantitative and Mixed Methods*. Sage; 2011.
- 460 40. National Institute for Health Research. The Research Cycle. [https://www.nihr.ac.uk/patients-
461 and-public/how-to-join-in/the-research-cycle/](https://www.nihr.ac.uk/patients-and-public/how-to-join-in/the-research-cycle/). Published 2018. Accessed June 21, 2018.
- 462 41. Ocloo J, Matthews R. From tokenism to empowerment: progressing patient and public
463 involvement in healthcare improvement. *BMJ Qual Saf*. 2016;25(March):1-7.
464 doi:10.1136/bmjqs-2015-004839.
- 465 42. Ennis L, Wykes T, Callaghan P, et al. Impact of patient involvement in mental health research:
466 longitudinal study. *Br J Psychiatry*. 2013;203(5):381-386. doi:10.1192/bjp.bp.112.119818.
- 467 43. PatientPartner. *Patient Involvement in Clinical Research: A Guide for Patient Organisations
468 and Patient Representatives*.; 2011.
469 <https://www.geneticalliance.org.uk/media/1602/patientpartnerforpatientorgs.pdf>. Accessed
470 February 20, 2018.
- 471 44. Greta Pecl, Chris Gillies, Carla Sbrocchi, Philip Roetman. *Building Australia Through Citizen
472 Science*.; 2015. [http://www.chiefscientist.gov.au/wp-content/uploads/Citizen-science-
473 OP_web.pdf](http://www.chiefscientist.gov.au/wp-content/uploads/Citizen-science-OP_web.pdf). Accessed August 24, 2017.
- 474 45. Lander J, Hainz T, Hirschberg I, Strech D. Current practice of public involvement activities in
475 biomedical research and innovation: A systematic qualitative review. *PLoS One*.
476 2014;9(12):e113274. doi:10.1371/journal.pone.0113274.
- 477 46. Rogers M, Bethel A, Boddy K. Development and testing of a medline search filter for
478 identifying patient and public involvement in health research. *Health Info Libr J*.
479 2017;34(2):125-133. doi:10.1111/hir.12157.

- 480 47. INVOLVE. *Guidance on Co-Producing a Research Project.*; 2018.
481 http://www.invo.org.uk/wp-content/uploads/2018/03/Copro_Guidance_Mar18.pdf. Accessed
482 March 14, 2018.
- 483 48. International Collaboration for Participatory Health Research (ICPHR). *Position Paper 1:*
484 *What Is Participatory Health Research? Version: May 2013.*; 2013.
485 [http://www.icphr.org/uploads/2/0/3/9/20399575/ichpr_position_paper_1_defintion_-](http://www.icphr.org/uploads/2/0/3/9/20399575/ichpr_position_paper_1_defintion_-_version_may_2013.pdf)
486 [_version_may_2013.pdf](http://www.icphr.org/uploads/2/0/3/9/20399575/ichpr_position_paper_1_defintion_-_version_may_2013.pdf). Accessed June 13, 2017.
- 487 49. Uher R. Genomics and the classification of mental illness: Focus on broader categories.
488 *Genome Med.* 2013. doi:10.1186/gm501.
- 489 50. Nunn JS, Scott CL, Stubbs JW, Cherry SF, Bismark MM. *Involving the Public in Rare Cancer*
490 *Care and Research.* Chichester, UK: John Wiley & Sons, Ltd; 2017:12-18.
491 doi:10.1002/9781119196235.ch3.
- 492 51. Haeusermann T, Greshake B, Blasimme A, Irdam D, Richards M, Vayena E. Open sharing of
493 genomic data: Who does it and why? *PLoS One.* 2017;12(5):e0177158.
494 doi:10.1371/journal.pone.0177158.
- 495 52. Messner DA, Koay P, Al Naber J, et al. Barriers to clinical adoption of nextgeneration
496 sequencing: A policy Delphi panel's solutions. *Per Med.* 2017;14(4):339-354.
497 doi:10.2217/pme-2016-0104.
- 498 53. Husedzinovic A, Ose D, Schickhardt C, Fröhling S, Winkler EC. Stakeholders' perspectives
499 on biobank-based genomic research: systematic review of the literature. *Eur J Hum Genet.*
500 2015;(January):1607-1614. doi:10.1038/ejhg.2015.27.